Differences in the Information Needs of Parents With a Child With a Genetic Syndrome:

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Differences in the information needs of parents with a child with a genetic syndrome: A cross-syndrome comparison

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Abstract

Background:

Due to the rarity of some genetic syndromes, information about these syndromes may be difficult for parents of affected children to access. Moreover, with specific behavioural phenotypes and these syndromes often being aggregated in large cohort studies, individual differences in informational needs and support across syndromes are not always reported.

Specific aims:

This study aimed to identify and contrast the most sought after information by parents' on the behavioural characteristics of three genetic syndromes: Cri du Chat (CdCS), Cornelia de Lange (CdLS) and Angelman syndromes (AS).

Method:

Ninety-eight parents (51 AS, 23 CdCS, 24 CdLS) completed an online survey that explored informational needs. Parents selected their three main informational needs of the past two years from a list of 32 topics.

Findings:

Communication, health and sleep were most frequently selected by parents of children with AS. In CdLS, behavioural changes with age, health and self-injury were selected, and in CdCS, health, behavioural changes with age and daily living skills. Significant differences in informational needs between the syndrome groups were found on the topics of behavioural changes with age, communication, autism spectrum disorder symptomatology, self-injury, and daily living skills.

Discussion:

The findings show that parents require a wide variety of information regarding their child's genetic syndrome and that the most sought after topics of information differ between syndromes. Therefore, it is important to avoid aggregating rare syndromes under broader

categories, as individual needs may be missed, and for policy and practice to take into consideration the differences in informational needs when tailoring support for families.

Differences in the information needs of parents with a child with a genetic syndrome: A cross-syndrome comparison

Rare genetic syndromes associated with intellectual disability (ID) are typically aggregated when examining priorities for support and research in the wider field of neurodevelopmental disorders (Allard et al., 2014; Morris et al., 2015). However, clinically significant differences in physical, cognitive, emotional and behavioural characteristics are observed in these syndromes (Arron, Oliver, Moss, Berg & Burbidge, 2011; Oliver, Berg, Moss, Arron & Burbidge, 2011). Such differences extend to parent variables, with levels of parental stress, for example, differentially elevated across groups (Hodapp, Wijma & Masino, 1997; Richman, Belmont, Kim, Slavin & Hayner, 2009; Wulffaert et al., 2009; Wulffaert, Scholte & van Berckelaer-Onnes, 2010). One aspect of parenting a child with a genetic syndrome that parents report increases stress is difficulty accessing information about their child's syndrome (Griffith et al., 2011). To date, there is limited research exploring cross syndrome differences in the information needs of parents of children with rare genetic syndromes. In this study, we explore these differences in a sample of syndromes; Cornelia de Lange (CdLS), Cri du Chat (CdCS) and Angelman syndromes (AS).

The European Commission defines a rare disease as affecting fewer than 5 in 10,000 individuals. 80% of rare diseases have a genetic origin (EURORDIS, 2007) with a significant number of genetic syndromes associated with intellectual disability (ID; Oliver, Woodcock & Adams, 2010). Although each rare disease affects a small proportion of people, collectively between 350,000 and 750,000 individuals in the United Kingdom are estimated to have a genetic disorder associated with ID (Oliver & Woodcock, 2008). The significant number of people who have a rare disease indicates a substantial need for advice and intervention (Dodge et al., 2010). A recent study has highlighted that a key priority of patients, carers and clinicians is further research in the field of neurodisability, which includes research into

optimal frequencies of mainstream therapies, the use of communication strategies and improving children's attitudes towards disabilities (Morris et al., 2015). However, this study aggregated conditions rather than focusing on individual disorders, which may mask priorities associated with rare genetic syndromes.

In the 2009 Annual Report, the Chief Medical Officer (CMO) discussed the urgent need to raise awareness and understanding of neglected rare diseases among professionals and the public. Subsequently, Rare Diseases UK (RDUK; 2011) placed a focus on the difficulties in accessing reliable, up-to-date information and the need for access to sufficient information at diagnosis and on an ongoing basis. In addition, inequalities in accessing information have been identified within the ID population (Emerson & Baines, 2011), thus compounding the problem for both rare diseases and ID.

This inability to access information for rare syndromes and ID has been shown to contribute to parental stress and impacts directly on parents' day-to-day lives (Griffith et al., 2011). In a qualitative study of parental experiences of support services for their child with CdCS, AS or CdLS, parents described having to 'battle' to get information and often feeling 'left in the dark' due to the rarity of their child's syndrome. They described how this situation consequently led to them feeling stressed and frustrated (Griffith et al., 2011). This lack of information was described alongside other issues including uneven medical and social care provision and the relentless need to be advocates for their child. These findings highlight that improved access to sufficient information that is tailored to parents' and/or syndrome-specific needs, could alleviate some of the stress experienced.

Further to this, there appears to be a discrepancy between the information parents want in regard to their child's syndrome and the information they are provided with. Hinkson and colleagues (2006) explored information preferences of parents with a child with CdLS. Reflux, behaviour and feeding problems were ranked as the highest three concerns of parents,

however information on reflux and behaviour was given to parents at diagnosis in less than half of cases. A bottom-up systematic approach of asking parents what information they require about their child's syndrome will allow practitioners to tailor the information they provide to parents to address their needs.

An important consideration is that needs of parents may differ based on the syndrome their child has. Differences in the phenotypes of the syndromes may influence the information that parents require. For example, clinically significant levels of overactivity and impulsivity are associated with CdCS and AS (Oliver et al., 2011) and individuals with CdLS have increased health problems compared to individuals with ID of heterogeneous aetiology (Hall, Arron, Sloneem & Oliver, 2008). These characteristics, among others associated with genetic syndromes, have an impact on the health and quality of life of individuals with a syndrome and their families and emphasises the need to look at these syndromes as individuals groups.

In summary, although individually rare, genetic syndromes associated with intellectual disability are cumulatively common (Dodge et al., 2010; Woodcock & Oliver, 2008). However, due to the rarity of individual syndromes information can be hard to access which can impact on parents' and carers' wellbeing (Griffith et al., 2011). Given these genetic syndromes evidence differing phenotypes, there is a need to look at parental needs for information at a cross-syndrome level to identify differences in these needs. Identifying these needs and differences will allow for resources and research dissemination to be tailored more appropriately which may contribute to improving parental wellbeing.

Therefore, the aim of this study is to identify the most important topics for parents with regard to the behavioural characteristics of their child with AS, CdCS or CdLS and to explore cross-syndrome differences in these informational needs.

Method

Participants

Parents were recruited in collaboration with the syndrome support groups: Angelman Syndrome Support, Education & Research Trust (ASSERT), Cornelia de Lange Syndrome Foundation UK & Ireland, Cri du Chat Syndrome Support Group who advertised the study through their mailing lists and social media pages. Participants were also recruited via email through a pre-existing database and social media page held by the Cerebra Centre for Neurodevelopmental Disorders (CCND). To ensure social media respondents from the CCND social media page were likely to be parents of children with a genetic syndrome they were asked to contact the Cerebra Centre to obtain a password to the survey. In total 98 parents completed the questionnaire (see Table 1 for demographic information). Due to the nature of recruitment, it was not possible to calculate a response rate as it is not known how many individuals received or had access to the call for participation.

Table 1
Demographic information for respondents of the questionnaire

	Syndromes		
	AS	CdCS	CdLS
Total number of informants	51	23	24
% mothers	86.3	91.3	87.5
% fathers	11.8	8.7	4.2
% long-term carers *(non-specified)*	1.9	0	8.3
Age of informant[1]	45.61 (9.36) 30.0-65.0	49.70 (12.24) 34.0-73.0	54.54 (7.79) 44.0-70.0
Age of person with genetic syndrome[1]	13.86 (10.22) 1.0-44.0	19.98 (11.93) 1.5-44.0	24.33(9.85) 7.0-46.0
Number of individuals with genetic syndromes who were over 18 (%)	27.5	56.5	79.2
% of those individuals who live at home	90.2	69.6	70.8

[1] *Mean (SD) and range.*

Measures

Demographic Questionnaire. The questionnaire included 14 items and covered information about the parent and their child with a genetic syndrome, including the demographic information presented in table 1.

Current Needs Checklist. An online checklist of informational needs was developed for the purpose of this study. Items on the checklist were selected after reviewing the literature on the support/clinical needs of individuals with intellectual disability and with data from a previous consultation exercise with parents of children with the syndromes of interest for another research project. This consultation was conducted at parent support group meetings and utilised focus groups and open-ended questions asking 'what information have you sought/or do you want about your child's syndrome'. This consultation exercise yielded responses from 60 individuals across six syndrome groups (fragile X, Prader-Willi, Smith-Magenis, Angelman, Cornelia de Lange and Cri du Chat syndromes). Three clinical psychologists who had regular clinical contact with individuals with intellectual disability and rare genetic syndromes reviewed a draft version of the checklist to ensure that, to the best of their knowledge, no topics typically raised through their clinical practice were overlooked.

Procedure

Participants were sent the link and password to access the online survey where they were asked to provide consent and fill out their demographic information. For the checklist, parents were asked to select the main three topics of information they had wanted in the past two years from a list of 32 topics. A full supplementary list of these topics is provided here [insert link]. These were presented in alphabetical order to facilitate respondents finding the most relevant topics and clear, simple terminology was used to faciliate understanding. To account for any topics that had been overlooked during the development of the checklist, an 'Other' option was included to allow parents to specify additional topics. The rationale for

limiting parents to the choice of three topics was that it would encourage parents to select the most important topics, allowing for comparisons across the groups.

Data Analysis

Due to the variability of participants' answers and to gain more statistical power, similar topics were collapsed prior to the analysis leaving 20 topics. '*Expressive communication, 'Receptive communication'* and *'Signing'* formed the *'Communication'* category, *'Toilet training', 'Washing/dressing'* and *'Mobility'* were collapsed into '*Daily living skills'* and *'Bone/joint problems', 'Cardiac problems', 'Dental problems', 'Ear/Eye infections', 'General health problems', 'Peripheral sensory neuropathy', 'Reflux'* and *'Seizures'* formed the *'Health'* category. *'Memory'* was merged with the *'Intellectual/Cognitive Characteristics'* category.

To identify parents' main informational needs, for each syndrome the proportion of the sample that selected a topic for each group was calculated. To ensure that proportions were not biased if informants selected more than one topic that was collapsed into a category, it was only counted once.

A one-way ANOVA was performed to explore whether the groups were matched for age prior to conducting the main analyses. There were significant differences in age, $F(2, 95) = 8.64$, $p < .001$, with Mann-Whitney U tests indicating that the AS group was younger than the CdLS and CdCS group, $U = 248$, $Z = 4.38$, $p < .001$ and $U = 389$, $Z = 2.31$, $p = .021$ respectively. There were no significant differences between CdLS and CdCS. As the AS group was significantly younger than the other groups, the groups were split into under and over 18s and Chi-square tests were employed to explore if there were any differences in the topics selected between younger and older samples in each group. No significant differences were found ($p > .05$) so no further age band splits were used in the main analyses.

When cells had fewer than five data points, a Fisher's exact analysis was used. As analysis was primarily exploratory, a less conservative *p*-value of .05 was used, as making a type-II error was deemed less preferable than missing clinical important differences through a type-I error.

Results

Descriptive statistics: Main information topics for parents

Of the original 32 topics, three topics (*Diagnosis procedure, Cardiac problems* and *Memory*) were not selected by any respondents. Once topics were condensed into the final 20 topics for analysis, *Diagnosis procedure* was the only topic not selected. The proportion of people selecting each of the 20 topics for each group is displayed in Table 2 along with the results of analyses.

For AS (N = 51), the topics selected by the highest percentage of individuals were *Health, Communication* and *Sleep*, with the percentage of all AS individuals selecting these topics: 59%, 41% and 26% respectively. For CdCS (N = 23) the main topics identified were *Health* (44%), *Behavioural changes with age* (39%), and *Daily living skills* (39%). For CdLS (N = 24) the topics selected were: *Behavioural changes with age* (58%), *Health* (50%), and *Self-injury* (25%).

Differences in topics selected between syndromes

Chi-square analysis showed significant differences between the three syndrome groups on the topics of *Behavioural changes with age*, X^2 (2, 98) = 8.76, p = .013, and *Communication* X^2 (2, 98) = 7.33, p = .026. Furthermore, post-hoc pairwise analysis showed that significantly more parents with a child with CdLS selected *Behavioural changes with age* than parents of children with AS, X^2 (1, 75) = 8.73, p = .003, and a significantly higher proportion of parents with children with AS selected the topic of *Communication* than parents with a child with CdLS, X^2 (1, 75) = 6.17, p = .013.

Fisher's exact analysis showed significant differences between the syndromes on four of the 20 topics. There were significant differences between parents of children with CdLS and parents of children with AS for the topic of *ASD characteristics (p* = .034, Fisher's exact test), with a higher proportion of parents of children with CdLS selecting this topic. Significantly more parents with children with CdCS selected the topic of *Self-injury* than parents of children with AS, (*p* = .006, Fisher's exact test), and differences between CdLS and AS for this topic were approaching significance, (*p* = .050, Fisher's exact test), with more parents of a child with CdLS choosing *Self-injury*. A significantly higher proportion of parents with children with CdCS selected the topic of *Daily living skills* than parents of children with CdLS (*p* = .038, Fisher's exact test). Finally, there were significant differences between parents of children with AS and parents of children with CdLS on the topic of sleep, with more parents with a child with AS selecting this topic, (*p* = .027, Fisher's exact test).

Table 2
Percentage of respondents selecting each topic on the questionnaire and statistical analysis showing the differences between the syndromes on these topics.

Topics of information	Syndrome group			X^2	*p*	Pairwise analyses[2]
	AS (N = 51)	CdCS (N = 23)	CdLS (N = 24)			
Aggression	15.7	13	12.5	-	-[1]	ns
ASD characteristics	2	8.7	16.7	-	-[1]	CdLS > AS
Behavioural changes with age	23.5	39.1	58.3	8.76	.013	CdLS > AS
Challenging behaviour: General	13.7	17.4	20.8	-	-[1]	ns
Communication	41.2	21.7	12.5	7.33	.026	AS > CdLS
Daily living skills	17.7	39.1	8.3	-	-[1]	CdCS > CdLS
Destructive/disruptive behaviour	9.8	4.3	0	-	-[1]	ns
Diagnosis procedure	0	0	0	-	-	-
Eating/feeding	9.8	17.4	8.3	-	-[1]	ns
Genetic mechanism	5.9	4.3	4.2	-	-[1]	ns
Health	58.8	43.5	50	1.62	.445	ns
Intellectual/cognitive characteristics	3.9	0	8.3	-	-[1]	ns
Mood and interest	3.9	8.7	4.2	-	-[1]	ns
Overactivity/impulsivity	0	4.3	0	-	-[1]	ns
Physical characteristics	0	4.3	0	-	-[1]	ns
Repetitive behaviour	3.9	0	0	-	-[1]	ns
Self-injury	7.8	34.8	25	-	-[1]	CdCS & CdLS > AS[3]
Sensory issues	11.8	0	8.3	-	-[1]	ns
Sleep	25.5	4.3	16.7	-	-[1]	AS > CdCS
Social behaviour	0	4.3	8.3	-	-[1]	ns
Other	9.8	8.6	8.3	-	-[1]	ns

[1] *Pairwise Fisher's exact tests conducted due to fewer than 5 data points in a group*
[2] $p < .05$
[3] $p < .01$
Note. A breakdown of percentages for all 31 topics included in the original checklist is available as supplementary online material.

Discussion

The aims of this study were to identify and compare the most frequently selected informational topics for parents with regard to the behavioural characteristics of their child's genetic syndrome. This aim was achieved via a checklist that was completed by parents who identified their top informational needs. There is limited literature directly comparing

informational needs of parents of children with different genetic syndromes. Studies that have explored informational needs have typically approached this at a broader level, whereas this study has allowed for a more in-depth investigation of needs associated with a diagnosis of genetic syndrome. The results of the current study suggest that policies or interventions that are generated based on aggregated cohorts may not be representative of individual syndrome groups within these cohorts.

Many of the results of this study suggest that parents' informational needs often align with the physical and behavioural characteristics of their child's genetic syndrome. For example, a frequently selected topic for CdCS and CdLS was *behavioural changes with age*. However, parents of individuals with AS were significantly less likely to endorse *behavioural changes with age* than parents of individuals with CdLS. These differences may be due to parental concerns about changes in behaviour that have been noted in adults with CdLS (Basile, Villa, Selicorni & Molteni, 2007; Kline et al., 2007; Sarimski, 1997). In AS, *communication* was endorsed more than in CdLS and *sleep* was endorsed more than in CdCS. Minimal or no speech and sleep difficulties are core characteristics and are included in the diagnostic criteria for AS (Williams et al., 2006). In contrast, in CdLS communication deficits are not as pervasive and seem to be associated with other characteristics of the syndrome (Ajmone et al., 2014; Goodban, 1993; Sarimski, 1997). Similarly in CdCS, sleep difficulties are no higher than in individuals with ID with a heterogeneous cause (Maas et al., 2009). A final example is the signifcantly lower numbers of parents of individuals with AS who selected *self-injury* in comparison to CdLS and CdCS. This complements research by Arron et al., (2011) who found that individuals with CdLS and CdCS were significantly more likely to show self-injurious behaviour than a control group of individuals with a heterogeneous cause of intellectual disability, whereas individuals with AS were no more likely to display this behaviour.

In busy health care environments, it is important to identify the key needs for families who approach services for support and advice about the developmental and health needs of their child with an intellectual disability. Research into the gaps in existing informational resources provides health care providers with knowledge of priority areas for the development of information. The current research suggests that the information that is most needed is often the information about the key characteristics of a specific disorder. Parents are not requiring information on obscure topics, but the topics that are central to supporting a person with that genetic syndrome. Providing this information is particularly important at the point where children are first diagnosed with an intellectual disability. When identifying key needs for children with ID it is appealing to conduct research at a broad level without consideration of the aetiology of a syndrome. This may be attractive given difficulties that arise with sampling from small populations and repeating studies across groups. However, for truly meaningful research that provides person centred informational resources, genetic aetiology is important (Arron, Oliver, Moss, Berg & Burbidge, 2011; Oliver, Berg, Moss, Arron & Burbidge, 2011). Health campaigns and written information provided by health care providers should take into consideration the importance of tailoring information to specific groups or, when this is not possible, at least acknowledging the importance of the characteristics of genetic syndromes in influencing the type of guidance that parents may need.

In recent years, there have been competing perspectives emerging in the literature with respect to the importance of genetic syndromes in determining approaches to health care. There is some concern that focusing on a person's genetic syndrome may lead to a belief in genetic determinism, which may distract from viewing persons with these syndromes as individuals (Waite et al., 2014). Another related concern, is genetic nihilism leading to interventions being denied to people with genetic syndromes due to a belief that

the behavioural characteristics of the syndrome are inevitable and unresponsive to intervention. At a similar time, there has been an increased emphasis on the importance of person centred care for individuals with ID (NICE, 2015). While there are dangers associated with an inflexible focus on the importance of someone's genetic syndrome, to be truly person centred we need to consider all aspects of the person in their care, and that means providing information that is tailored to the characteristics of a population. To simply aggregate cohorts into generalised ID, may reduce the nuances and mean that information is not meeting needs effectively. The importance of focusing on genetic aetiology of ID was explored recently via interviews with parents of individuals with genetic syndromes and professionals (Redley, Pannebaker and Holland, 2016). Parents' and professionals' accounts suggested that there are benefits in the medicalisation of genetic syndromes and incorporating the genetic knowledge into the provision of health and social care for individuals with a genetic syndrome and their families.

Alongside topics selected most frequently relating to key characteristics of the syndromes, other frequent topics were related to factors that have been identified as contributing to parental stress. For example challenging behaviour, which includes self-injury, contributes to parental stress in CdLS (Richman et al., 2009; Sarimski, 1997) and lower adaptive skills in individuals with CdCS have been suggested as a contributing factor to parental stress (Hodapp et al., 1997). Further research needs to be conducted on this possible association, and it may be that by providing information on topics that contribute to parental stress, parental well-being could be increased.

An alternative perspective is that the information parents' had sought over the last two years, may not reflect the information that has the most benefit these parents or their children. It is possible that, while families may seek information on specific syndrome related difficulties, information seeking may be driven by parental variables, such as anxiety and

intolerance of uncertainty; variables that have been shown to influence information seeking (Rosen & Knäuper, 2009). If this were evidenced, tackling parental responses to having a child diagnosed with a rare genetic syndrome using acceptance-based interventions may be more beneficial to parents in the long-term. For example, acceptance and commitment therapy has been shown to improve parents' well-being (MacDonald et al., 2010). Policy makers and clinicians should be mindful that improving outcomes for parents and their children is likely to rely on a combination of developing syndrome specific resources and information, and working with families around their perceptions of their child's difficulties.

A number of limitations to this current study must be considered. Firstly, there are two limitations of the checklist. The checklist did not include descriptors of each category. These were omitted as most categories did not require further explanation (e.g. sleep) and it was felt that descriptors would lengthen the measure making it less accessible to parents. However, there is a possibility that parents may have interpreted the categories inconsistently. While inconsistency in interpretation may reduce the likelihood of detecting significant differences, this is unlikely to explain the systematic differences observed across the groups. A secondary limitation, is that no psychometric testing of the checklist was conducted. As the reliability was not established, the claims of the study are preliminary and should be used to guide further research into these topics. However, due to the lack of research in this area and specificity of this study, there was not a previously validated measure that could be utilised.

Secondly, for the Chi-square analyses a less conservative *p* value of .05 was used, increasing the probability of a type I error. However, due to small samples and the importance of not overlooking clinically relevant differences, it is more appropriate to follow up findings than completely overlook potential differences. Whilst the lack of adjustment of the *p* value increased the likelihood of finding cross-syndrome differences by chance, the

pattern of differences in this study were in concordance with the previously reported behavioural phenotypes of each syndrome; thus, the results have face validity and suggest that exploring the differences in information needs based on genetic syndrome may be important.

Another limitation with regard to the sample is that the children of parents in the AS group were significantly younger than those in the CdLS and CdCS groups. However, age group splits followed by within-group age analysis showed that there were no significant differences between the topics chosen by parents of individuals over the age of 18 and those under 18 for any of the syndromes. Therefore, this goes some way to suggesting that age differences were not a confounding factor in determining the topics parents selected. Nevertheless, future research should confirm this by looking at younger samples for CdLS and CdCS.

Finally, it is possible that there is a selection bias in the sample and that the sample may not be representative of the population of parents of children with these genetic conditions. Further research needs to be conducted to expand these samples and to understand further the group differences that have been observed in this study. Despite this, this does not invalidate the main finding of this study that differences in parents' informational needs are likely to exist across syndromes.

These findings highlight that the information needs of parents correspond to factors that contribute to parental stress in these syndromes and behaviours that are associated with the syndrome's behavioural phenotype. This suggests that research needs to carry on investigating these areas to develop greater understanding but most importantly focus on disseminating findings to parents and tailoring resources to the individual syndromes. Future research exploring the information needs of parents should look at the topics in more detail,

perhaps incorporating a qualitative approach. Identifying specific aspects of topics will contribute to directing research in these areas.

To summarise, this research shows that there are differences in the information needs of parents of children with different genetic syndromes. Therefore, it is important to look at these syndromes at a cross-syndrome level in order to avoid the needs of parents being missed due to the rarity of these syndromes.

References

Allard, A., Fellowes, A., Shilling, V., Janssens, A., Beresford, B., & Morris, C. (2014). Key health outcomes for children and young people with neurodisability: qualitative research with young people and parents. *BMJ open*, *4*(4), e004611. https://doi.org/10.1136/bmjopen-2013-004611

Ajmone, P. F., Rigamonti, C., Dall'Ara, F., Monti, F., Vizziello, P., Milani, D., ... & Costantino, A. (2014). Communication, cognitive development and behavior in children with Cornelia de Lange syndrome (CdLS): preliminary results. *American Journal of Medical Genetics Part B: Neuropsychiatric Genetics*, *165*(3), 223-229. http://dx.doi.org/10.1002/ajmg.b.32224

Arron, K., Oliver, C., Moss, J., Berg, K., & Burbidge, C. (2011). The prevalence and phenomenology of self-injurious and aggressive behaviour in genetic syndromes. *Journal of Intellectual Disability Research*, *55*, 109–120. http://dx.doi.org/10.1111/j.1365-2788.2010.01337.x

Basile, E., Villa. L., Selicorni, A., & Molteni, M. (2007). The behavioural phenotype of Cornelia de Lange syndrome: a study of 56 individuals. *Journal of Intellectual Disability Research*, *51*(9), 671-68. http://dx.doi.org/10.1111/j.1365-2788.2007.00977.x

Chief Medical Officer. (2010). *2009 Annual Report of the Chief Medical Officer*. Retrieved from: http://www.sthc.co.uk/Documents/CMO_Report_2009.pdf. Accessed 25/08/2016

Dodge, J. A., Chigladze, T., Donadieu, J., Grossman, Z., Ramos, F., Serlicorni, A., … Wierzba, J. (2011). The importance of rare diseases: from the gene to society. *Archives of Disease in Childhood*, *96*(9), 791-792. http://dx.doi.org/10.1136/adc.2010.193664

Emerson, E., & Baines, S. (2011). Health inequalities and people with learning disabilities in the UK. *Tizard Learning Disability Review*, *16*(1), 42-48.

EURORDIS. (2007). *What is a rare disease?* Retrieved from: http://www.eurordis.org/sites/default/files/publications/Fact_Sheet_RD.pdf. Accessed 25/08/2016

Goodban, M. T. (1993). Survey of speech and language skills with prognostic indicators in 116 patients with Cornelia de Lange syndrome. *American Journal of Medical Genetics*, *47*(7), 1059-1063. http://dx.doi.org/10.1002/ajmg.1320470725

Griffith, G. M., Hastings, R. P., Nash, S., Petalas, M., Oliver, C., Howlin, P., … Tunnicliffe, P. (2011). "You have to sit and explain it all, and explain yourself." Mothers' experiences of support services for their offspring with a rare genetic intellectual disability syndrome. *Journal of Genetic Counseling*, *20*(2), 165–177. http://dx.doi.org/10.1007/s10897-010-9339-4

Hall, S. S., Arron, K., Sloneem, J., & Oliver, C. (2008). Health and sleep problems in Cornelia de Lange Syndrome: a case control study. *Journal of Intellectual Disability Research*, *52*(5), 458–468. http://dx.doi.org/10.1111/j.1365-2788.2008.01047.x

Hinkson, D. A. R., Atenafu, E., Kennedy, S. J., Vohra, S., Garg, D., & Levin, A. V. (2006). Cornelia de Lange syndrome: Parental preferences regarding the provision of medical information. *American Journal of Medical Genetics Part A*, *140A*(20), 2170–2179. http://dx.doi.org/10.1002/ajmg.a.31404

Hodapp, R. M. (1997). Direct and indirect behavioral effects of different genetic disorders of mental retardation. *American Journal on Mental Retardation*, *102*(1), 67–79. http://dx.doi.org/10.1352/0895-8017(1997)102%3C0067:DAIBEO%3E2.0.CO;2

Hodapp, R. M., Wijma, C. A., & Masino, L. L. (1997). Families of children with 5p- (cri du chat) syndrome: familial stress and sibling reactions. *Developmental Medicine and Child Neurology*, *39*(11), 757–761.

Kline, A. D., Krantz, I. D., Sommer, A., Kliewer, M., Jackson, L. G., FitzPatrick, D. R., ... & Selicorni, A. (2007). Cornelia de Lange syndrome: clinical review, diagnostic and scoring systems, and anticipatory guidance. *American Journal of Medical Genetics Part A*, *143*(12), 1287-1296. http://dx.doi.org/10.1002/ajmg.a.31757

Maas, A. P. H. M., Didden, R., Korzilius, H. P. L. M., Braam, W., Smits, M. G., & Curfs, L. M. G. (2009). Sleep in individuals with Cri du Chat syndrome: a comparative study. *Journal of Intellectual Disability Research*, *53*(8), 704-715. http://dx.doi.org/10.1111/j.1365-2788.2009.01184.x

MacDonald, E. E., & Hastings, R. P. (2010). Mindful parenting and care involvement of fathers of children with intellectual disabilities. *Journal of Child and Family Studies*, *19*(2), 236-240. https://doi.org/10.1007/s10826-008-9243-9

Morris, C., Simkiss, D., Busk, M., Morris, M., Allard, A., Denness, J., ... & Cowan, K. (2015). Setting research priorities to improve the health of children and young people with neurodisability: a British Academy of Childhood Disability-James Lind Alliance Research Priority Setting Partnership. *BMJ open*, *5*(1), e006233. http://dx.doi.org/10.1136/bmjopen-2014-006233

Oliver, C., Berg, K., Moss, J., Arron, K., & Burbidge, C. (2011). Delineation of behavioral phenotypes in genetic syndromes: Characteristics of Autism Spectrum Disorder, affect and hyperactivity. *Journal of Autism and Developmental Disorders*, *41*(8), 1019–1032. http://dx.doi.org/10.1007/s10803-010-1125-5

Oliver, C., & Woodcock, K. (2008). Integrating levels of explanation in behavioural phenotype research. *Journal of Intellectual Disability Research*, *52*, 807–809. DOI: 10.1111/j.1365-2788.2008.01117.x

Oliver, C., Woodcock, K. & Adams, D. (2010). *The importance of aetiology of intellectual disability.* In: Grant, G., Ramcharan, P., Flynn, M. and Richardson, M. (Eds.) Learning Disability: A life cycle approach to valuing people. (pp. 135-146). Open University Press\Wiley.

Rare Diseases UK. (2011). *Improving Lives, Optimising Resources: A Vision for the UK Rare Disease Strategy.* Retrieved from: http://www.raredisease.org.uk/documents/RD-UK-Strategy-Report.pdf.

Redley, M., Pannebakker, M., & Holland, A. (2016). Improving the Health and Well- Being of Adults With Conditions of a Genetic Origin: Views from Professionals, Syndrome Support Groups and Parents. *Journal of Applied Research in Intellectual Disabilities*. http://dx.doi.org/10.1111/jar.12293

Richman, D. M., Belmont, J. M., Kim, M., Slavin, C. B., & Hayner, A. K. (2009). Parenting stress in families of children with Cornelia de Lange Syndrome and Down Syndrome. *Journal of Developmental and Physical Disabilities*, *21*(6), 537–553. http://dx.doi.org/10.1007/s10882-009-9156-6

Rosen, N. O., & Knäuper, B. (2009). A little uncertainty goes a long way: State and trait differences in uncertainty interact to increase information seeking but also increase worry. *Health communication*, *24*(3), 228-238. http://dx.doi.org/10.1080/10410230902804125

Sarimski, K. (1997). Communication, social-emotional development and parenting stress in Cornelia-de-Lange syndrome. *Journal of Intellectual Disability Research*, *41*(1), 70-75. DOI: 10.1111/j.1365-2788.1997.tb00678.x

Waite, J., Heald, M., Wilde, L., Woodcock, K., Welham, A., Adams, D., & Oliver, C. (2014). The importance of understanding the behavioural phenotypes of genetic syndromes associated with intellectual disability. *Paediatrics and Child Health*, *24*(10), 468-472. http://dx.doi.org/10.1016/j.paed.2014.05.002

Williams, C. A., Beaudet, A. L., Clayton-Smith, J., Knoll, J. H., Kyllerman, M., Laan, L. A., ... & Wagstaff, J. (2006). Angelman syndrome 2005: updated consensus for diagnostic criteria. *American Journal of Medical Genetics Part A*,*140*(5), 413-418. http://dx.doi.org/10.1002/ajmg.a.31074

Wulffaert, J., van Berckelaer-Onnes, I., Kroonenberg, P., Scholte, E., Bhuiyan, Z., & Hennekam, R. (2009). Simultaneous analysis of the behavioural phenotype, physical factors, and parenting stress in people with Cornelia de Lange syndrome. *Journal of Intellectual Disability Research*, *53*(7), 604–619. http://dx.doi.org/10.1111/j.1365-2788.2009.01185.x

Wulffaert, J., Scholte, E. M., & Van Berckelaer-Onnes, I. A. (2010). Maternal parenting stress in families with a child with Angelman syndrome or Prader-Willi syndrome. *Journal of Intellectual & Developmental Disability*, *35*(3), 165–174. http://dx.doi.org/10.3109/13668250.2010.499101

Supplementary Table

Percentage of respondents selecting each topic on the questionnaire and statistical analysis showing the differences between the syndromes on these topics.

Topics of information	Syndrome group			X^2	*p*	Pairwise analyses[2]
	AS (N = 51)	CdCS (N = 23)	CdLS (N = 24)			
Aggression	15.7	13	12.5	-	-[1]	ns
ASD characteristics	2	8.7	16.7	-	-[1]	CdLS > AS
Behavioural changes with age	23.5	39.1	58.3	8.76	.013	CdLS > AS
Challenging behaviour: General	13.7	17.4	20.8	-	-[1]	ns
Communication:	41.2	21.7	12.5	7.33	.026	AS > CdLS
Expressive	25.5	13	4.2	-	-[1]	AS > CdLS
Receptive	15.7	4.3	8.3	-	-[1]	ns
Signing	7.8	4.3	0	-	-[1]	ns
Daily Living Skills:	17.7	39.1	8.3	-	-[1]	CdCS > CdLS
Mobility	17.6	21.7	4.2	-	-[1]	ns
Toilet Training	3.9	8.7	4.2	-	-[1]	ns
Washing/dressing	0	13	4.2	-	-[1]	ns
Destructive/disruptive behaviour	9.8	4.3	0	-	-[1]	ns
Diagnosis procedure	0	0	0	-	-	-
Eating/feeding	9.8	17.4	8.3	-	-[1]	ns
Genetic mechanism	5.9	4.3	4.2	-	-[1]	ns
Health:	58.8	43.5	50	1.62	.445	ns
Bone/joint problems	9.8	17.4	4.2	-	-[1]	ns
Cardiac problems	0	0	0	-	-	-
Dental problems	5.9	13	12.5	-	-[1]	ns
Ear/Eye infections	3.9	0	4.2	-	-[1]	ns
General problems	11.8	13	12.5	-	-[1]	ns
Peripheral sensory neuropathy	0	0	4.2	-	-[1]	ns
Reflux	11.8	8.7	25	-	-[1]	ns
Seizures	33.3	0	0	-	-	AS > CdCS & CdLS[4]
Intellectual/cognitive characteristics:	3.9	0	8.3	-	-[1]	ns
Memory	0	0	0	-	-	-
Mood and interest	3.9	8.7	4.2	-	-[1]	ns
Overactivity/impulsivity	0	4.3	0	-	-[1]	ns
Physical characteristics	0	4.3	0	-	-[1]	ns
Repetitive behaviour	3.9	0	0	-	-[1]	ns
Self-injury	7.8	34.8	25	-	-[1]	CdCS & CdLS > AS[3]
Sensory issues	11.8	0	8.3	-	-[1]	ns
Sleep	25.5	4.3	16.7	-	-[1]	AS > CdCS
Social behaviour	0	4.3	8.3	-	-[1]	ns
Other	9.8	8.6	8.3	-	-[1]	ns

[1] *Pairwise Fisher's exact tests conducted due to fewer than 5 data points in a group*

[2] $p < .05$, [3] $p < .01$, [4] $p < .001$